1. SHOCK

Child with prolonged Capillary Filling time (CFT), [Normal is <3 secs)

Age	Lower Limit of Normal Systolic Blood Pressure
Term neonate (0 to 28 days)	More than 60 mm Hg or strong central pulse
Infant (1 to 12 months)	More than 70 mm Hg or strong central pulse
Child 1 to 10 years	More than 70 + (2 × age in years)
Child 10 years or older	More than 90 mm Hg

Neonates: Umbilical pulse

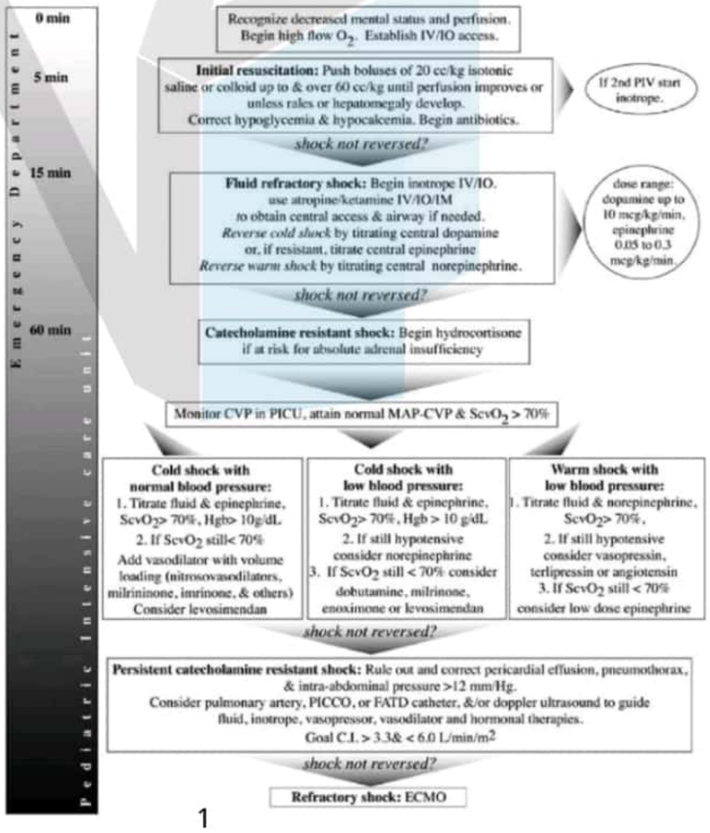
Normal Saline Bolus

- Newborn = 10mL/kg
- Child = 20mL/kg
- Max= 60 mL/kg

➢ Cold Shock: Nor-Epinephrine

➢ Warm Shock: Epinephrine

➢ Newborns: Dopamine



One Liners- Pediatric Shock

1. Fluid of choice for Pediatric Shock — Normal Saline
2. IV antibiotics within how many minutes — 60 min
3. Target ScvO2>? — >70%
4. Inotrope in Cold Shock? — epinephrine
5. Inotrope in Warm Shock? — norepinephrine
6. Steroids role? — IV hydrocortisone

2. A Choking Infant

Foreign bodies common in children

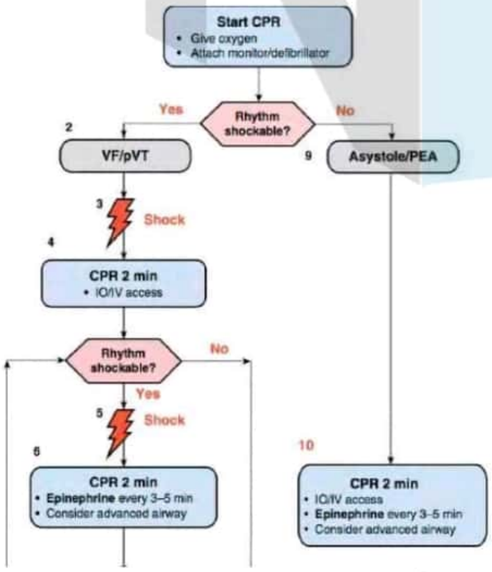
Infants: Back slaps & Chest thrusts

Age group- 1 to 3 years

M/C FB: Nuts

Alternate between 5 Back slaps and 5 chest thrusts until F.B. dislodges.

3. Pediatric Cardiac Arrest

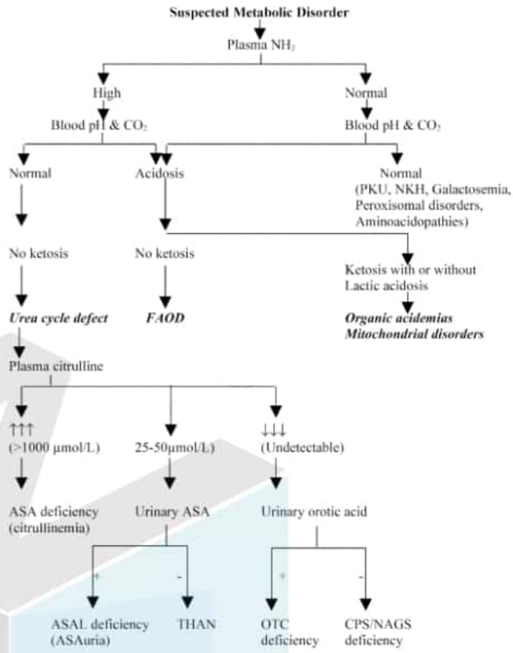


Most common cause: Asystole

4. Infant IEM Approach

Hints:

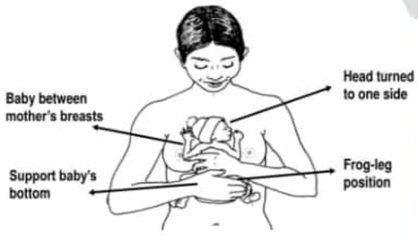
- Born Normal
- Unexplained lethargy
- No signs of sepsis
- Abnormal Urine odour
- Family history of consanguinity



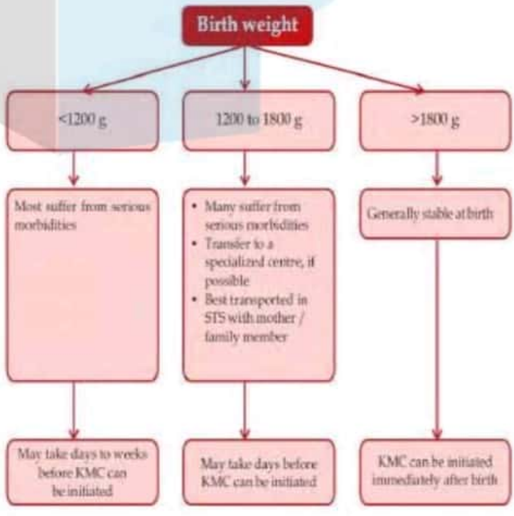
- **In urea cycle d/o, sometimes respiratory alkalosis can also be seen.**
- Organic Acidemia + Rash→ Multiple Carboxylase deficiency
  Rx: Biotin
- **M/C Urea Cycle d/o: OTC deficiency**
- FAOD: Fatty Acid Oxidation defect

5. Low Birth Weight

KMC



- To prevent hypothermia
- Started when baby's weight >1800g


Birth weight
<1200 g
1200 to 1800 g
>1800 g
Most suffer from serious morbidities
Many suffer from serious morbidities
Transfer to a specialized centre, if possible
Best transported in STS with mother / family member
Generally stable at birth
May take days to weeks before KMC can be initiated
May take days before KMC can be initiated
KMC can be initiated immediately after birth

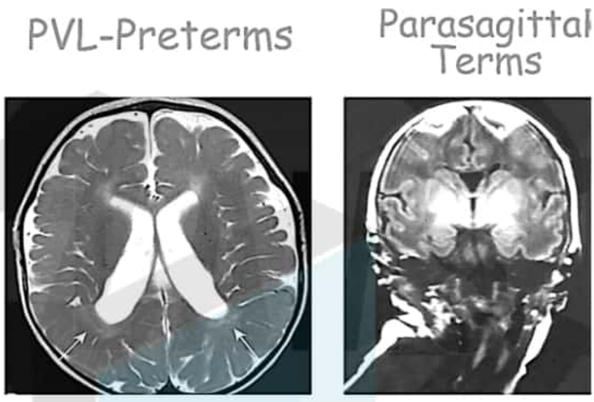
One Liners- thermoregulation

1. Delivery room temperature- 25 C (WHO)
2. Mechanism Incubator & wraps→ Reduce convection
3. Mechanism- Radiant warmer, Radiant heat
4. KMC initiated at what weight? →All LBWs
5. KMC stopped at? →2500gms

6. Asphyxial injury in Newborns

PVL: Periventricular Leukomalacia
(Less white mater around ventricles)
C/F: Spastic Diplegia later in life
Seen in Pre-terms
Scissoring gait

Para-Sagittal insults
(Involvement of Basal Ganglia)
C/F- Quadriplegia later in life
Term babies



T2 weighted MRI Image

Rx: THERAPEUTIC HYPOTHERMIA

- Cool the head or whole body
- Temp: 33.5 degree Celsius
- Should be started within 6 hours of birth
- Should Continue till 72 hrs
- Reduces the free radical injury to the brain

7,8. Preterm with Respiratory distress

C/F : GRUNTING (expiratory sound showing breathing against closed glottis)

FEATURE	SCORE 0	SCORE 1	SCORE 2
Chest Movement	Equal	Respiratory Lag	Seesaw Respiration
Intercostal Retraction	None	Minimal	Marked
Xiphoid Retraction	None	Minimal	Marked
Nasal Flaring	None	Minimal	Marked
Expiratory Grunt	None	Audible w/ stethoscope	Audible

SILVERMAN ANDERSON RETRACTION SCORE

>4 → Distress

>7 → Severe distress (Impending Respi Failure)

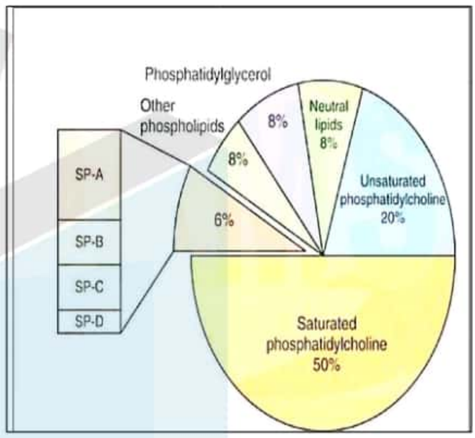
Downes' score

Score	Respiratory rate	Cyanosis	Air entry	Grunt	Retraction
0	<60/min	Nil	Normal	None	Nil
1	60- 80/min	In room air	Mild ?	Ausc with stethoscope	Mild
2	>80/min	present	Marked ?	Audible with naked ear	Moderate

- Mild: 0-3, moderate: 4-6 , severe: 7-10
- A score of >6 is indicative of impending respiratory failure.

COMPOSITION OF SURFACTANT →

- Surfactant is formed by Type II Pneumocytes
- Formed at about 20 weeks IUL
- Formed in Canalicular phase of lung development



Treatment of RDS:

Early use of nCPAP for stabilization of at-risk preterm infants beginning early (in the delivery room) reduces the need for mechanical ventilation.

Assisted ventilation and surfactant are indicated for infants with RDS who cannot keep oxygen saturation >90% while breathing 40–70% oxygen and receiving nCPAP.

substantial evidence supports the feasibility and efficacy of prophylactic nCPAP as the *primary* means of respiratory support for preterm infants with RDS. CPAP started at birth is as effective as prophylactic or early surfactant and is associated with a reduction in BPD. *Prophylactic nCPAP is therefore the approach of choice for the delivery room management of a preterm neonate at risk for RDS.*

RDS Rx

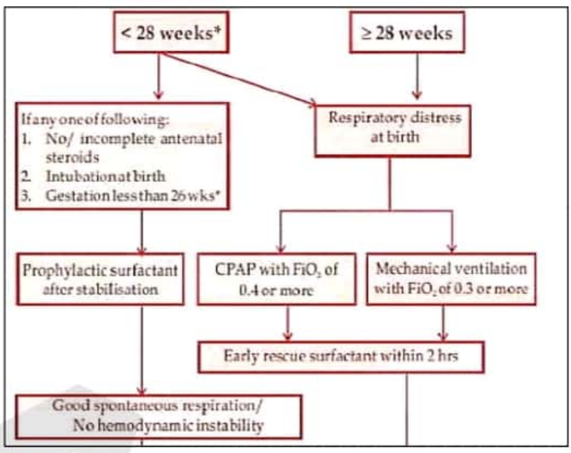
- Minimum 100 mg/kg surfactant
- Rapid bolus technique
- InSurE

IN tubation SUR factant Rapid Extubation

Newer Technique: LISA → Less Invasive Surfactant Administration (via Forceps and NG Tube) (No intubation Required)

5

ONE LINERS ON RDS


< 28 weeks*
≥ 28 weeks
If any one of following:
1. No/ incomplete antenatal steroids
2. Intubation at birth
3. Gestation less than 26 wks*
Respiratory distress at birth
Prophylactic surfactant after stabilisation
CPAP with FiO_2 of 0.4 or more
Mechanical ventilation with FiO_2 of 0.3 or more
Early rescue surfactant within 2 hrs
Good spontaneous respiration/ No hemodynamic instability

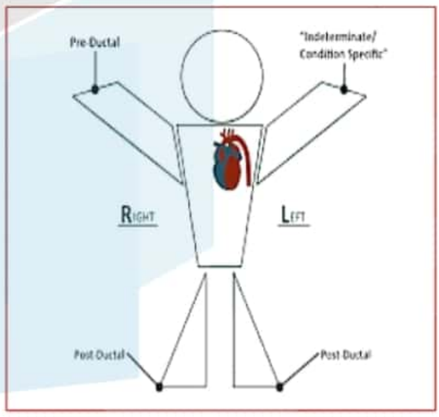
1. 1st therapy in delivery room for VLBW → Ncpap
2. Target SpO2 preterm? → 91-95% (Hyperoxia can cause Retrolental Fibroplasia)
3. Most common tachypnea newborn → TTNB
4. Baby not vigorous meconium stained liquor? →PPV-
   If Tone, Respiratory Effort and HR>100bpm you can directly go for PPV in MSL.

9. New NALS 2015 Recommendations

Q. Which limb is used for SpO2 monitoring in a newborn
A. RIGHT UPPER LIMB

Q. What is 3-minute target for spO2 in newborns?
A. 70-75%

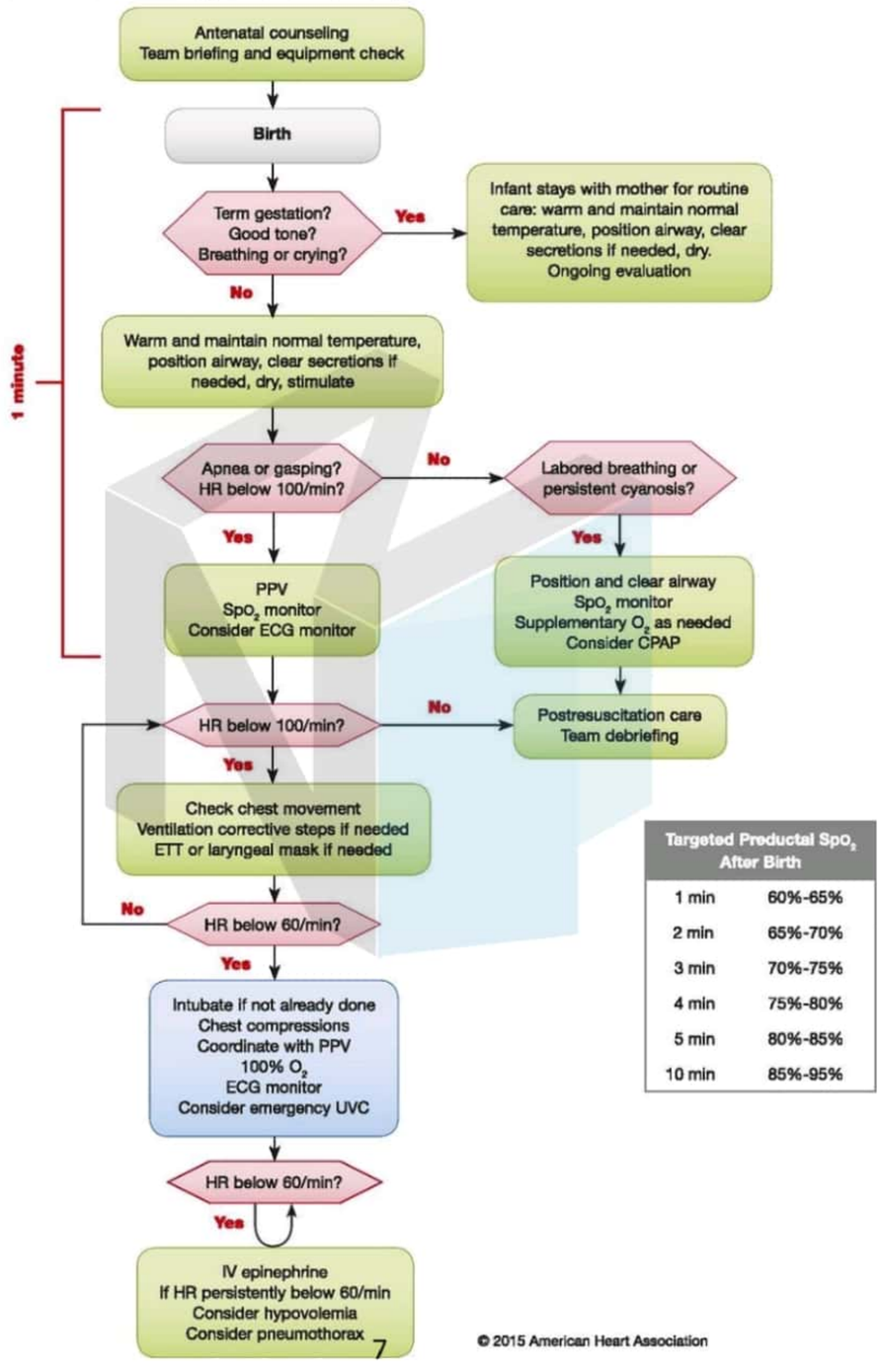
Targeted Preductal SpO_2 After Birth	
1 min	60%-65%
2 min	65%-70%
3 min	70%-75%
4 min	75%-80%
5 min	80%-85%
10 min	85%-95%



Important Points:

- While Suctioning, Mouth is suctions first followed by nose.
- TABC: Temperature, Airway, Breathing, Circulation
- UVC: Umbilical Vein Catheter
- Epinephrine Dose: 0.1 to 0.3 mL/kg intravenously in 1:10000 dilution
  Nearly equivalent to 0.01 -0.03 mg/kg
- Intratracheal Epinephrine can also be given alternatively, dose is 10 times than that of IV.

Neonatal Resuscitation Algorithm—2015 Update



10. Preterm with abdominal distention

Dx: Necrotising Enterocolitis

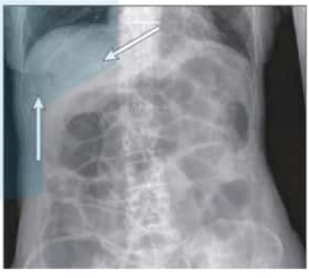
MODIFIED BELL's STAGING CRITERIA

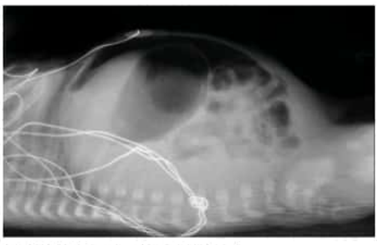
Stage	Systemic signs	Abdominal signs	Radiographic signs	Treatment
IA Suspected	Temperature instability, apnea, bradycardia, lethargy	Gastric retention, abdominal distention, emesis, heme-positive stool	Normal or intestinal dilation, mild ileus	NPO, antibiotics x 3 days
IB Suspected	Same as above	Grossly bloody stool	Same as above	Same as IA
IIA Definite, mildly ill	Same as above	Same as above, plus absent bowel sounds with or without abdominal tenderness	Intestinal dilation, ileus, pneumatosis intestinalis	NPO, antibiotics x 7 to 10 days
IIB Definite, moderately ill	HYPONATREMIA Same as above, plus mild metabolic acidosis and thrombocytopenia	Same as above, plus absent bowel sounds, definite tenderness, with or without abdominal cellulitis or right lower quadrant mass	Same as IIA, plus ascites Portal Venous Gas	NPO, antibiotics x 14 days
IIIA Advanced, severely ill, intact bowel	Same as IIB, plus hypotension, bradycardia, severe apnea, combined respiratory and metabolic acidosis, DIC, and neutropenia	Same as above, plus signs of peritonitis, marked tenderness, and abdominal distention	Same as IIA, plus ascites	NPO, antibiotics x 14 days, fluid resuscitation, inotropic support, ventilator therapy, paracentesis
IIIB Advanced, severely ill, perforated bowel	Same as IIIA	Same as IIIA	Same as above, plus pneumoperitoneum	Same as IIA, plus surgery

DIC: disseminated intravascular coagulation
NPO: "nil per os" or nothing by mouth

Portal Venous Gas

Arrow indicates portal venous gas (Air in the right hypochondrium)





PNEUMOPERITONEUM

In a Cross Table Lateral XRAY

Pneumatosis Intestinalis

Arrow indicates intramural gas in bowel wall

8

11. Newborn with Seizure

TYPES OF NEONATAL SEIZURES

Four types of neonatal seizures
1. Subtle seizures
2. Clonic seizures
3. Tonic seizures
4. Myoclonic seizures

SUBTLE SEIZURES

- Most common form(>50%)
- It includes

a) Ocular - tonic horizontal deviation of eyes or sustained eye opening with ocular fixation or cycled fluttering.
b) Oral facial lingual movements - chewing, tongue thrusting, lip smacking etc.
c) Limb movements - cycling, paddling, boxing etc.
d) Autonomic phenomena-tachycardia or bradycardia.
e) Apnea may be a rare manifestation of seizure.

CLONIC SEIZURES

- Rhythmic movements of muscle groups.
- Have both fast and slow movements with frequency of 1-3 jerks per second.
- Commonly associated with EEG changes.
- May be unifocal or multifocal.
- Focal clonic has good prognosis.

TONIC SEIZURES

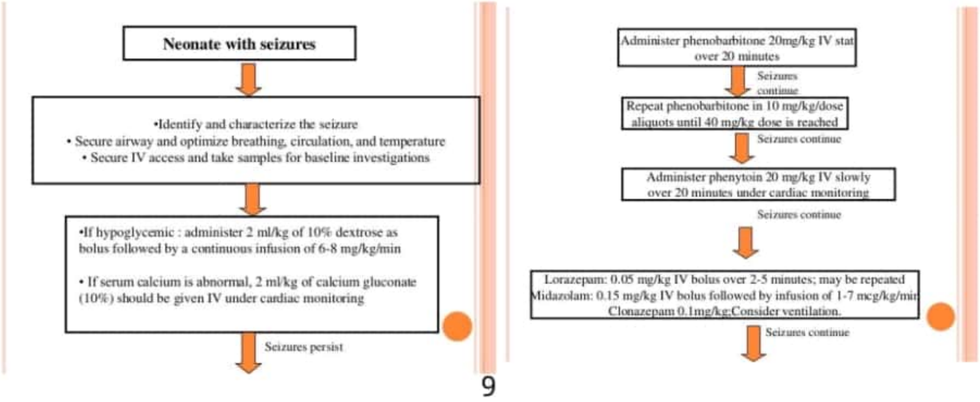
- Pattern is sustained posture of limbs or asymmetrical truncal postures.
- cause: diffuse neurological injury or IVH in preterm or postasphyxial.
- Usually no EEG changes.
- Prognosis is poor except for postasphyxial cases.

MYOCLONIC SEIZURES

- Non rhythmic lightning fast contraction.
- Seen in diffuse brain damage as in perinatal asphyxia, inborn errors of metabolism, cerebral dysgenesis.
- Worst prognosis in terms of neurodevelopmental outcome and seizure recurrence.

NEONATAL SEIZURES – TIME OF ONSET

Time of onset	Etiology
< 24 hrs	HIE, severe birth trauma, hypoglycemia, hypocalcemia, drug withdrawl, congenital CNS anomalies, intracranial hemorrhage
1-3 days	All above , subarachnoid hemorrhage, IEM, benign familial neonatal seizures
> 3 days	sepsis ,meningitis, progressive hydrocephalus, epileptic syndromes, herpes encephalitis, IEM


Neonate with seizures
•Identify and characterize the seizure
• Secure airway and optimize breathing, circulation, and temperature
• Secure IV access and take samples for baseline investigations
•If hypoglycemic : administer 2 ml/kg of 10% dextrose as bolus followed by a continuous infusion of 6-8 mg/kg/min
• If serum calcium is abnormal, 2 ml/kg of calcium gluconate (10%) should be given IV under cardiac monitoring
Seizures persist
Administer phenobarbitone 20mg/kg IV stat over 20 minutes
Seizures continue
Repeat phenobarbitone in 10 mg/kg/dose aliquots until 40 mg/kg dose is reached
Seizures continue
Administer phenytoin 20 mg/kg IV slowly over 20 minutes under cardiac monitoring
Seizures continue
Lorazepam: 0.05 mg/kg IV bolus over 2-5 minutes; may be repeated
Midazolam: 0.15 mg/kg IV bolus followed by infusion of 1-7 mcg/kg/min
Clonazepam 0.1mg/kg;Consider ventilation.
Seizures continue

Second line drugs like
Lidocaine[4mg/kg f/b 2mg/kg/hr]
Paraldehyde[0.1-0.2ml/kg/dose IM]
sodium valproate[20-25mg/kg f/b 5-10mg/kg/12h]
Topiramate(20mg/kg/day)
Levetiracetam(10-30mg/kg/day)
Vigabatrin(50mg/kg/day) Pyridoxine(100mgIVtestdose)
exchange transfusion[IEMs,drug toxicity,bilirubin encephalopathy

Seizures controlled

Wean AEDs slowly to maintenance phenobarbitone

12. NEONATAL REFLEXES (Illingworth)

	Appearance	Disappearance	Remarks
Moro reflex	The Moro reflex is a vestibular reflex that is present in incomplete form in premature birth after the 28th week of gestation, and is usually present in complete form by week 34 (third trimester) peaks in the first month of life.	Begins to disappear by 2 months of age but can be noted till 6 months of age.	An absent or inadequate Moro response on one side is found in infants with hemiplegia, brachial plexus palsy, or a fractured clavicle/humerus. Cannot be elicited in severe tone abnormalities. Persistence of the Moro response beyond 4 or 5 months of age is noted only in infants with severe neurological defects. When eliciting the response, the head should be in the midline and the hands should be open.
Asymmetric tonic neck reflex	Onset: 0–2 months	Normally vanishes around 6 months of age.	Functional significance: persistence of this reflex may indicate CNS damage. The ATNR and tonic labyrinthine reflex (TLR) both hinder functional activities such as rolling, bringing the hands together, or even bringing the hands to the mouth. Over time, both the ATNR and TLR can cause serious damage to the growing child's joints and bones. The ATNR can cause the spine to curve (scoliosis). Both the ATNR and TLR can cause the head of the thighbone to partially slip out (subluxation) or completely move out of the hip socket (dislocation).
Symmetric tonic neck reflex	Onset: 4–6 months	Integration: 8–12 months	Functional significance: necessary to achieve quadruped crawling.
Grasp reflex Palmar grasp Plantar grasp	These two grasp reflexes appear at birth.	It should have disappeared in 2 or 3 months; while the palmar grasp fades at 5 or 6 months of age, the plantar remains during the first year.	Functional significance:following the development of grasp, the infant begins to reach for objects and utilises a crude palmar grasp to hold them. Persistence may indicate the spastic form of cerebral palsy.
Landau reflex	Onset: 3–4 months	Integration 1 year	Functional significance: breaks up the total flexion pattern seen at birth. Absence of the reflex over the age of 3 months is seen in cases of motor weakness, cerebral palsy and intellectual disability.

13. Milestones

15 MO
Motor: Walks alone; crawls up stairs
Adaptive: Makes tower of 3 cubes; makes a line with crayon; inserts raisin in bottle
Language: Jargon; follows simple commands; may name a familiar object (e.g., ball); responds to his/her name
Social: Indicates some desires or needs by pointing; hugs parents

18 MO
Motor: Runs stiffly; sits on small chair; walks up stairs with 1 hand held; explores drawers and wastebaskets
Adaptive: Makes tower of 4 cubes; imitates scribbling; imitates vertical stroke; dumps raisin from bottle
Language: 10 words (average); names pictures; identifies 1 or more parts of body
Social: Feeds self; seeks help when in trouble; may complain when wet or soiled; kisses parent with pucker

24 MO
Motor: Runs well, walks up and down stairs, 1 step at a time; opens doors; climbs on furniture; jumps
Adaptive: Makes tower of 7 cubes (6 at 21 mo); scribbles in circular pattern; imitates horizontal stroke; folds paper once imitatively
Language: Puts 3 words together (subject, verb, object)
Social: Handles spoon well; often tells about immediate experiences; helps to undress; listens to stories when shown pictures

30 MO
Motor: Goes up stairs alternating feet
Adaptive: Makes tower of 9 cubes; makes vertical and horizontal strokes, but generally will not join them to make cross; imitates circular stroke, forming closed figure
Language: Refers to self by pronoun "I"; knows full name
Social: Helps put things away; pretends in play

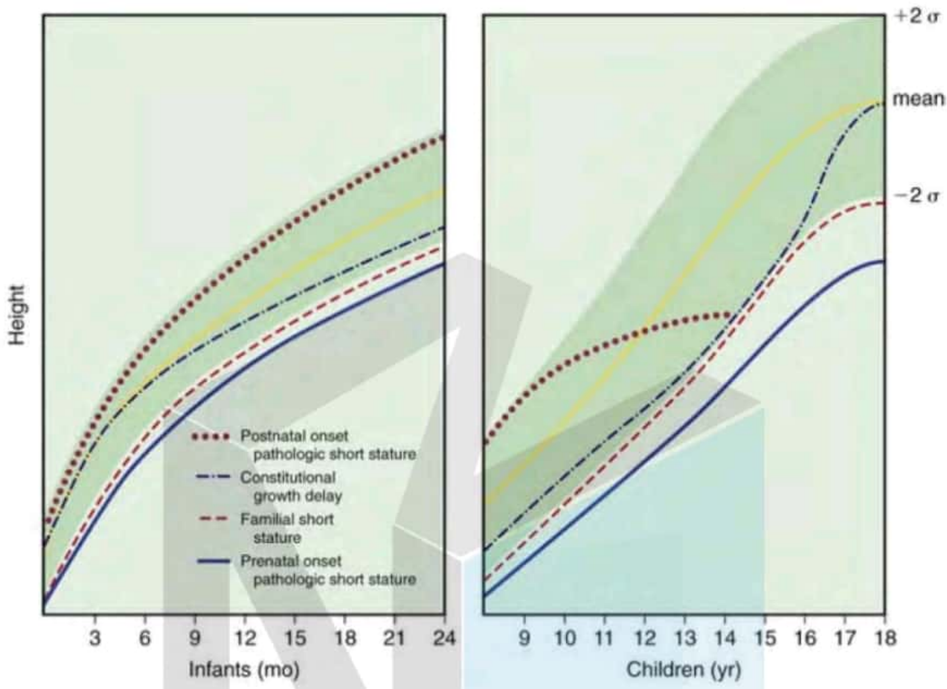
36 MO
Motor: Rides tricycle; stands momentarily on 1 foot
Adaptive: Makes tower of 10 cubes; imitates construction of "bridge" of 3 cubes; copies circle; imitates cross
Language: Knows age and sex; counts 3 objects correctly; repeats 3 numbers or a sentence of 6 syllables; most of speech intelligible to strangers
Social: Plays simple games (in "parallel" with other children); helps in dressing (unbuttons clothing and puts on shoes); washes hands

48 MO
Motor: Hops on 1 foot; throws ball overhand; uses scissors to cut out pictures; climbs well
Adaptive: Copies bridge from model; imitates construction of "gate" of 5 cubes; copies cross and square; draws man with 2-4 parts besides head; identifies longer of 2 lines
Language: Counts 4 pennies accurately; tells story
Social: Plays with several children, with beginning of social interaction and role-playing; goes to toilet alone

60 MO
Motor: Skips
Adaptive: Draws triangle from copy; names heavier of 2 weights
Language: Names 4 colors; repeats sentence of 10 syllables; counts 10 pennies correctly
Social: Dresses and undresses; asks questions about meaning of words; engages in domestic role-playing

14. SHORT STATURE

→ Most Common Cause: Constitutional



15,16 Pedigree Chart Analysis

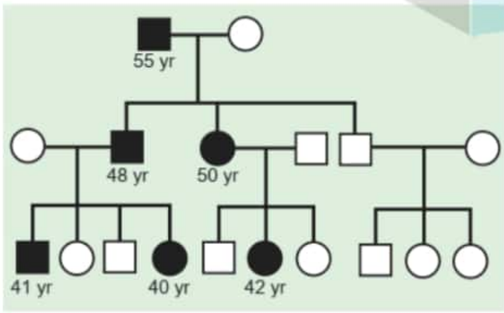
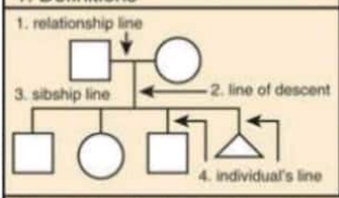
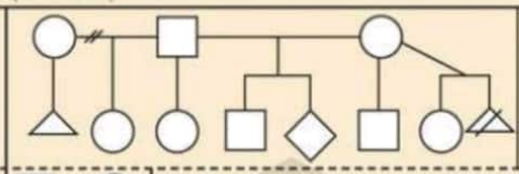
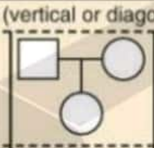
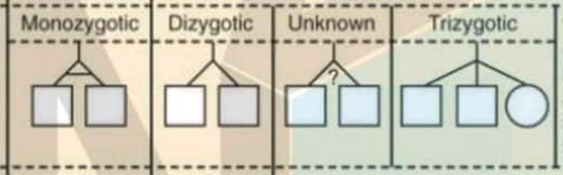
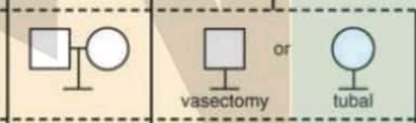
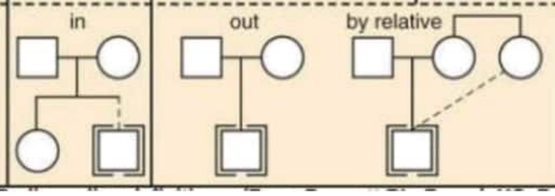


Figure 80-16 Myotonic dystrophy pedigree illustrating anticipation. In this case, the age at onset for family members affected with an autosomal dominant disease is lower in more recent generations. *Black*, affected patients.

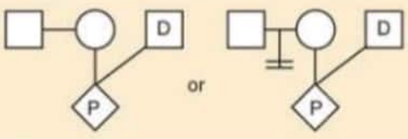
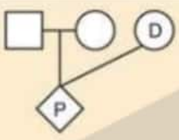
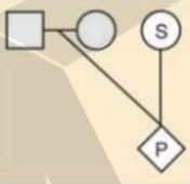
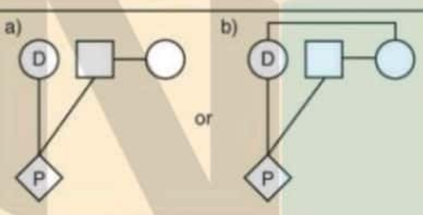
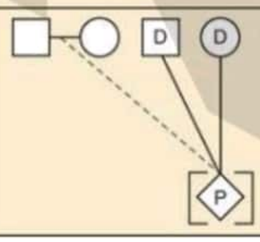
Instructions:
—Key should contain all information relevant to interpretation of pedigree (e.g., define fill/shading)
—For clinical (non-published) pedigrees include:
a) name of proband/consultand
b) family names/initials of relatives for identification, as appropriate
c) name and title of person recording pedigree
d) historian (person relaying family history information)
e) date of intake/update
f) reason for taking pedigree (e.g., abnormal ultrasound, familial cancer, developmental delay, etc.)
g) ancestry of both sides of family
—Recommended order of information placed below symbol (or to lower right)
a) age; can note year of birth (e.g., b.1978) and/or death (e.g., d. 2007)
b) evaluation (see Figure 75-4)
c) pedigree number (e.g., I-1, I-2, I-3)
—Limit identifying information to maintain confidentiality and privacy

	Male	Female	Gender not specified	Comments
1. Individual	b.1925	30 y	4 mo	Assign gender by phenotype (see text for disorders of sex development, etc.). Do not write age in symbol.
2. Affected individual				Key/legend used to define shading or other fill (e.g., hatches, dots, etc.). Use only when individual is clinically affected.
				With ≥2 conditions, the individual's symbol can be partitioned accordingly, each segment shaded with a different fill and defined in legend.
3. Multiple individuals, number known	5	5	5	Number of siblings written inside symbol. (Affected individuals should not be grouped.)
4. Multiple individuals, number unknown or unstated	n	n	n	"n" used in place of "?".
5. Deceased individual	d. 35	d. 4 mo	d. 60's	Indicate cause of death if known. Do not use a cross (†) to indicate death to avoid confusion with evaluation positive (+).
6. Consultand				Individual(s) seeking genetic counseling/testing.
7. Proband	P	P		An affected family member coming to medical attention independent of other family members.
8. Stillbirth (SB)	SB 28 wk	SB 30 wk	SB 34 wk	Include gestational age and karyotype, if known.
9. Pregnancy (P)	P LMP: 7/1/2007 47,XY,+21	P 20 wk 46, XX	P	Gestational age and karyotype below symbol. Light shading can be used for affected; define in key/legend.

Pregnancies not carried to term	Affected	Unaffected	
10. Spontaneous abortion (SAB)	17 wks female cystic hygroma	<10 wks	If gestational age/gender known, write below symbol. Key/legend used to define shading.
11. Termination of pregnancy (TOP)	18 wks 47, XY, +18		Other abbreviations (e.g., TAB, VTOP) not used for sake of consistency.
12. Ectopic pregnancy (ECT)	ECT		Write ECT below symbol.

1. Definitions	Comments
	If possible, male partner should be to left of female partner on relationship line. Siblings should be listed from left to right in birth order (oldest to youngest).
2. Relationship line (horizontal)	
a. Relationships 	A break in a relationship line indicates the relationship no longer exists. Multiple previous partners do not need to be shown if they do not affect genetic assessment.
b. Consanguinity	If degree of relationship not obvious from pedigree, it should be stated (e.g., third cousins) above relationship line.
3. Line of descent (vertical or diagonal)	
a. Genetic 	Biologic parents shown.
- Multiple gestation 	The horizontal line indicating monozygosity is placed between the individual's line and not between each symbol. An asterisk (*) can be used if zygosity proven.
- Family history not available/ known for individual	
- No children by choice or reason unknown 	Indicate reason, if known.
- Infertility	Indicate reason, if known.
b. Adoption 	Brackets used for all adoptions. Adoptive and biological parents denoted by dashed and solid lines of descent, respectively.

Instructions:
— D represents egg or sperm donor
— S represents surrogate (gestational carrier)
— If the woman is both the ovum donor and a surrogate, in the interest of genetic assessment, she will only be referred to as a donor (e.g., 4 and 5); the pregnancy symbol and its line of descent are positioned below the woman who is carrying the pregnancy
— Available family history should be noted on the gamete donor and/or gestational carrier

Possible Reproductive Scenarios		Comments
1. Sperm donor		Couple in which woman is carrying pregnancy using donor sperm. No relationship line is shown between the woman carrying the pregnancy and the sperm donor.
2. Ovum donor		Couple in which woman is carrying pregnancy using a donor egg and partner's sperm. The line of descent from the birth mother is solid because there is a biologic relationship that may affect the fetus (e.g., teratogens).
3. Surrogate only		Couple whose gametes are used to impregnate a woman (surrogate) who carries the pregnancy. The line of descent from the surrogate is solid because there is a biological relationship that may affect the fetus (e.g., teratogens).
4. Surrogate ovum donor		Couple in which male partner's sperm is used to inseminate (a) an unrelated woman or (b) a sister who is carrying the pregnancy for the couple.
5. Planned adoption		Couple contracts with a woman to carry a pregnancy using ovum of the woman carrying the pregnancy and donor sperm.

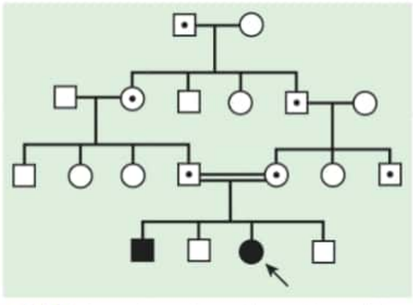


Figure 80-7 Autosomal recessive pedigree with parental consanguinity. *Central dot*, carriers; *black*, affected patients.

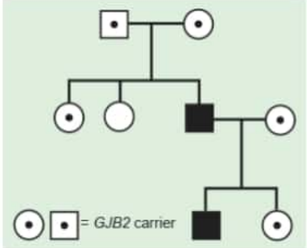


Figure 80-8 Pseudodominant inheritance. *Black*, affected (deaf); *central dot* shows carrier who is asymptomatic (unaffected).

Instructions:
— E is used for evaluation to represent clinical and/or test information on the pedigree
  a. E is to be defined in key/legend
  b. If more than one evaluation, use subscript (E_1, E_2, E_3) and define in key
  c. Test results should be put in parentheses or defined in key/legend
— A symbol is shaded only when an individual is clinically symptomatic
— For linkage studies, haplotype information is written below the individual. The haplotype of interest should be on left and appropriately highlighted
  Repetitive sequences, trinucleotides, and expansion numbers are written with affected allele first and placed in parentheses
— If mutation known, identify in parentheses

Definition	Symbol	Scenario
1. Documented evaluation (*) Use only if examined/evaluated by you or your research/clinical team or if the outside evaluation has been reviewed and verified.	*	Woman with negative echocardiogram. * E– (echo)
2. Carrier—not likely to manifest disease regardless of inheritance pattern		Male carrier of Tay-Sachs disease by patient report (* not used because results not verified).
3. Asymptomatic/presymptomatic carrier—clinically unaffected at this time but could later exhibit symptoms		Woman age 25 with negative mammogram and positive BRCA1 DNA test. * 25 y E_1– (mammogram) E_2+ (5385insC BRCA1)
4. Uninformative study (u)	Eu	Man age 25 with normal physical exam and uninformative DNA test for Huntington disease (E_2). * 25 y E_1– (physical exam) E_2u (36n/18n)
5. Affected individual with positive evaluation (E+)	E+	Individual with cystic fibrosis and positive mutation study; only one mutation has currently been identified. E+(ΔF508) Eu * E+(ΔF508/u)
		10 week male fetus with a trisomy 18 karyotype. P * 10 wk E+(CVS) 47,XY,+18

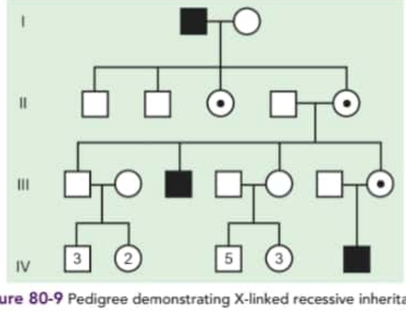


Figure 80-9 Pedigree demonstrating X-linked recessive inheritance.

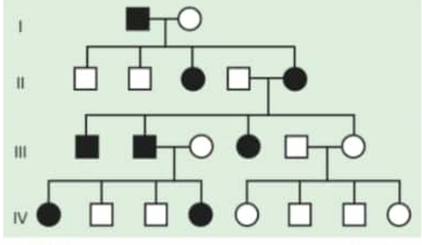


Figure 80-11 Pedigree pattern demonstrating X-linked dominant inheritance. Note there is no father-to-son transmission in this situation, and hemizygosity (i.e., X-linked gene in a male) is not lethal. In some X-linked dominant conditions, X-linked males have a more severe phenotype and might not survive. In that case, only females manifest the disease (see Fig. 80-12).

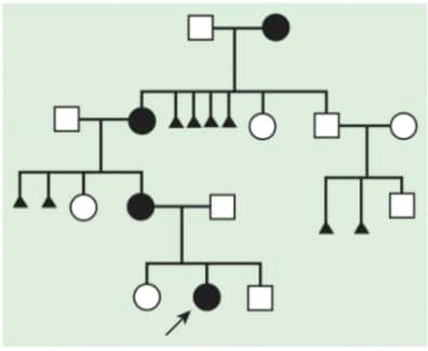


Figure 80-12 Pedigree of an X-linked dominant disorder with male lethality, such as incontinentia pigmenti.

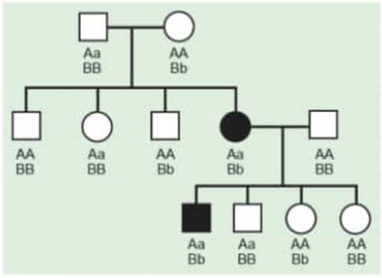


Figure 80-14 Digenic pedigree. Here, the disease alleles are *a* and *b* and they reside on distinct genetic loci or genes. For a person to have the disease, heterozygosity for mutant alleles in both genes (A/a;B/b) is required.

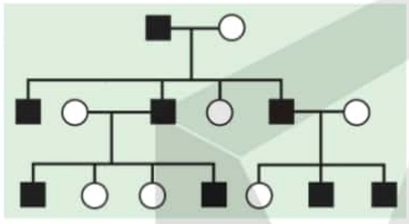


Figure 80-13 Y-linked inheritance. *Black*, affected patient.

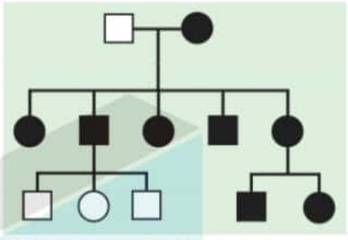


Figure 80-15 Pedigree of a mitochondrial disorder, exhibiting maternal inheritance. *Black*, affected patient.

Table 80-1 Representative Examples of Disorders Caused by Mutations in Mitochondrial DNA and Their Inheritance

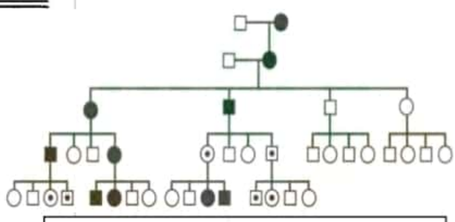
DISEASE	PHENOTYPE	MOST FREQUENT MUTATION IN MTDNA MOLECULE	HOMOPLASMY VS. HETEROPLASMY	INHERITANCE
Leber hereditary optic neuropathy	Rapid optic nerve death, leading to blindness in young adult life	Substitution Arg340His in *ND1* gene of complex I of electron transport chain; other complex I missense mutations	Homoplasmic (usually)	Maternal
NARP, Leigh disease	Neuropathy, ataxia, retinitis pigmentosa, developmental delay, mental retardation, lactic acidemia	Point mutations in ATPase subunit 6 gene	Heteroplasmic	Maternal
MELAS	Mitochondrial encephalomyopathy, lactic acidosis, and strokelike episodes; may manifest only as diabetes mellitus	Point mutation in $tRNA^{Leu}$	Heteroplasmic	Maternal
MERRF	Myoclonic epilepsy, ragged red fibers in muscle, ataxia, sensorineural deafness	Point mutation in $tRNA^{Lys}$	Heteroplasmic	Maternal
Deafness	Progressive sensorineural deafness, often induced by aminoglycoside antibiotics	A1555G mutation in 12S rRNA	Homoplasmic	Maternal
	Nonsyndromic sensorineural deafness	A7445G mutation in 12S rRNA	Homoplasmic	Maternal
Chronic progressive external ophthalmoplegia (CPEO)	Progressive weakness of extraocular muscles	The common MELAS point mutation in $tRNA^{Lys}$; large deletions similar to KSS	Heteroplasmic	Maternal if point mutations
Pearson syndrome	Pancreatic insufficiency, pancytopenia, lactic acidosis	Large deletions	Heteroplasmic	Sporadic, somatic mutations
Kearns-Sayre syndrome (KSS)	PEO of early onset with heart block, retinal pigmentation	5 kb large deletion	Heteroplasmic	Sporadic, somatic mutations

mtDNA, Mitochondrial DNA; rRNA, ribosomal RNA; tRNA, transfer RNA.
From Nussbaum RL, McInnes RR, Willard HF, editors: Thompson & Thompson genetics in medicine, *ed 6, Philadelphia, 2001, WB Saunders, p. 246.*

Genomic Imprinting disorders

Table 1. Principal Features of Prader–Willi Syndrome and Angelman Syndrome.

Prader–Willi Syndrome	Angelman Syndrome
Mild or moderate mental retardation	Severe mental retardation
Feeding problems	Ataxia
Neonatal and infantile hypotonia	Hyperactivity
Hyperphagia	Convulsions
Obesity	Paroxysms of laughter
Genital hypoplasia	Almost-absent speech
Characteristic facies	Clumsy, jerky movements
Dolichocephaly	Characteristic facies
Narrow bifrontal diameter	Microcephaly
Almond-shaped eyes	Prognathism (progenia)
Small-appearing mouth	Macrostomia (large mouth)
Down-turned corners of the mouth	Tongue protrusion
Behavioral problems	Widely spaced teeth
Short stature	Ocular hypopigmentation
Small hands and feet	Abnormal electroencephalogram
Hypopigmentation	



Angelman Syndrome Pedigree (15q11-13)

Most common genetic cause of obesity: Prader-willi Synd

Prader Willi Syndrome

Table 81-20 Molecular Mechanisms Causing Prader-Willi and Angelman Syndromes

	PRADER-WILLI SYNDROME	ANGELMAN SYNDROME
15q11-q13 deletion	~70% (paternal)	~70% (maternal)
Uniparental disomy	~30% (maternal)	~5% (paternal)
Single-gene mutation	None detected	E6-AP ubiquitin-protein ligase (10% of total but seen only in familial cases)
Imprinting center mutation	5%	5%
Unidentified	<1%	10-15%

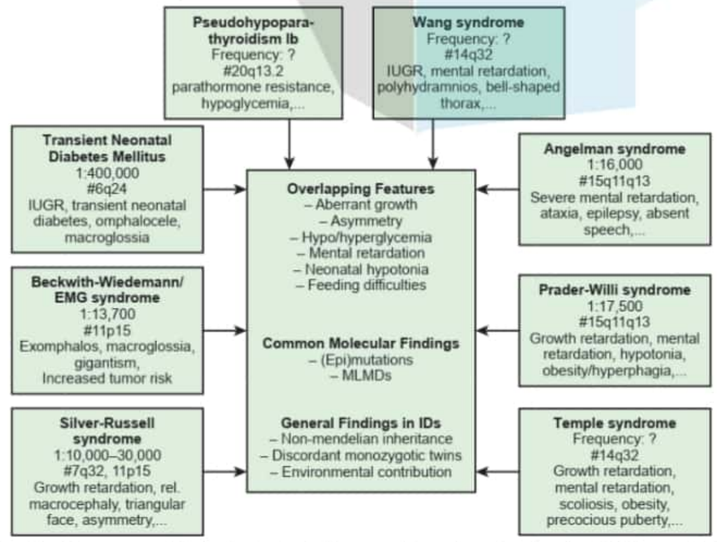
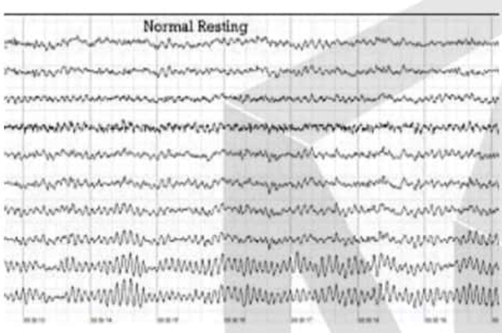


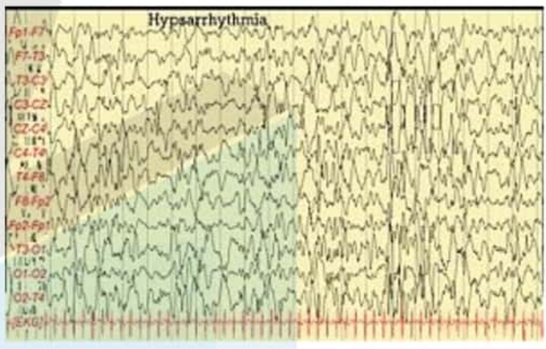
Figure 81-18 Overview of the common clinical and molecular findings in the 8 known imprinting disorders, including specific features, frequencies, and major chromosomal localization. *#6q24*, chromosome 6q24; *EMG*, exomphalos-macroglossia-gigantism; *IUGR*, intrauterine growth restriction. *(From Eggermann T, Elbracht M, Schröder C, et al: Congenital imprinting disorders: a novel mechanism linking seemingly unrelated disorders. J Pediatr 163:1202-1207, 2013, Fig. 2, p. 1204).*

SYSTEMIC PEDIATRICS

Case 17: Infantile Spasm/West Syndrome/Salaam Seizure

- Salaam Seizure: B/L Symmetric Flexion movement
- Seen in between 4-8 months
- EEG: Generalized Chaotic Pattern **(HYPSARRYTHMIA)**
- Triad:
  - Infantile spams
  - Hypsarrythmia
  - Developmental retardation/regression
- During spasm- EEG slows (Electro-decremental response)





Case 18: Absence seizure

The hallmark of the absence seizures is abrupt and sudden-onset impairment of consciousness, interruption of ongoing activities, a blank stare, possibly a brief upward rotation of the eyes. If the patient is speaking, speech is slowed or interrupted; if walking, they stand transfixed; if eating, the food will stop on its way to the mouth. Usually, the patient will be unresponsive when addressed. The attack lasts from a few seconds to half a minute and evaporates as rapidly as it commenced. Absence seizures generally are not followed by a period of disorientation or lethargy (post-ictal state)

Table 7. **Comparison of Typical and Atypical Absence Seizures**

Factor	Typical	Atypical
Age of onset	Childhood	Any age
Onset/offset of seizure	Abrupt	Often gradual
Consciousness	Totally lost	Often partially impaired
Other clinical features during seizure	Slight (eye flickering)	Can be prominent, including aura, automatism
Duration of seizures	Short (usually <10 sec)	Long (usually several minutes)
Frequency of seizures	Numerous, frequently in clusters	Usually less frequent
Postictal	None	Confusion, headache, emotional disturbance are common
Coexisting seizure types	Sometimes tonic-clonic and myoclonic	Mixed seizure disorder is common; all seizure types
Cause	Idiopathic generalized epilepsy	Any focal pathology or probably symptomatic epilepsy
Underlying focal anatomic lesion	None	Limbic structures, neocortex
Other neurologic signs and symptoms	None	Usually learning difficulties
Ictal EEG appearance	3-Hz spike and wave	2 to 2.5-Hz spike and wave
Interictal EEG appearance	Usually normal	Abnormal

- **Atypical absence seizures :**
- Associated myoclonic components.
- Tone changes of the head (head drop) and body.
- Variable impairments of consciousness.
- Last longer than typical absences.
- Precipitated by drowsiness.
- Not provoked by hyperventilation or photic stimulation.
- Usually more difficult to treat.

Rx: DOC for-

Typical Sz- ETHOSUXIMIDE

Atypical Sz- VALPROATE

- **1-2 Hz spike–and–slow-wave discharges.**

Juvenile Myoclonic Epilepsy

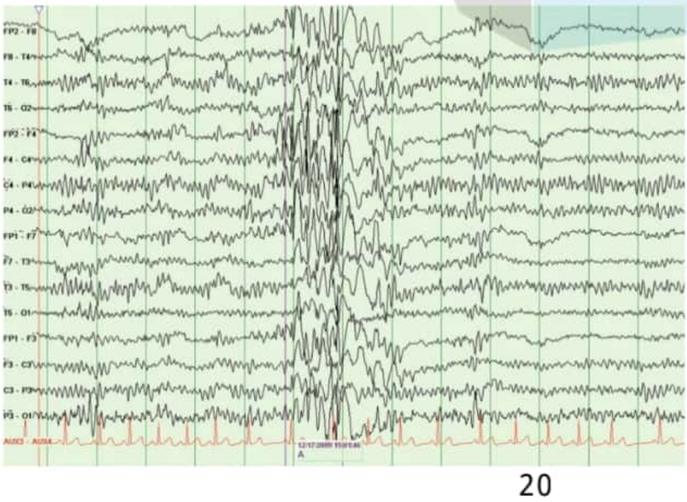
- Most common among IGEs: 4-6%
- Genetically determined
- 40-50 %: family history of epilepsy
- Myoclonic seizures (MSs): 100%
- Generalized tonic-clonic seizures: 90%
- Absence seizures: 35%
- EEG-3-6 Hz spike/polyspike-slow waves with intradischarge fragmentation and unstable frequency
- One third of patients have photoparoxysmal responses

58

JME is the most common epilepsy in adoloscents and young adults

Aggrevating factor: Light

Therefore mostly happen in early morning when sunlight appears



Polyspike & wave discharges
Stimulated by photic stimulations

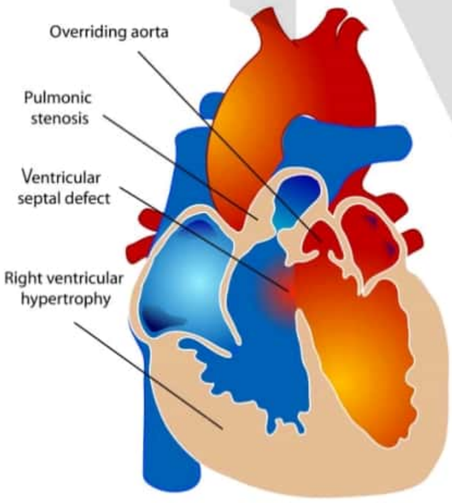
Rx:
- DOC- Valproate
- For women- Levetiracetam (Due to hormonal and fetal side effects of Valproate)
- Children- Levetiracetam
- Other drugs: Lamotrigine, Zonisamide, Topiramate, Perampanel

Case 19: Congenital Heart Disease

Table 424-1	Relative Frequency of Major Congenital Heart Lesions*
LESION	**% OF ALL LESIONS**
Ventricular septal defect	35-30
Atrial septal defect (secundum)	6-8
Patent ductus arteriosus	6-8
Coarctation of aorta	5-7
Tetralogy of Fallot	5-7
Pulmonary valve stenosis	5-7
Aortic valve stenosis	4-7
D-Transposition of great arteries	3-5
Hypoplastic left ventricle	1-3
Hypoplastic right ventricle	1-3
Truncus arteriosus	1-2
Total anomalous pulmonary venous return	1-2
Tricuspid atresia	1-2
Single ventricle	1-2
Double-outlet right ventricle	1-2
Others	5-10

Most Common **Cyanotic** Heart Disease→ Tetralogy of Fallot

Tetralogy of Fallot



Radiology in Tetralogy of Fallot

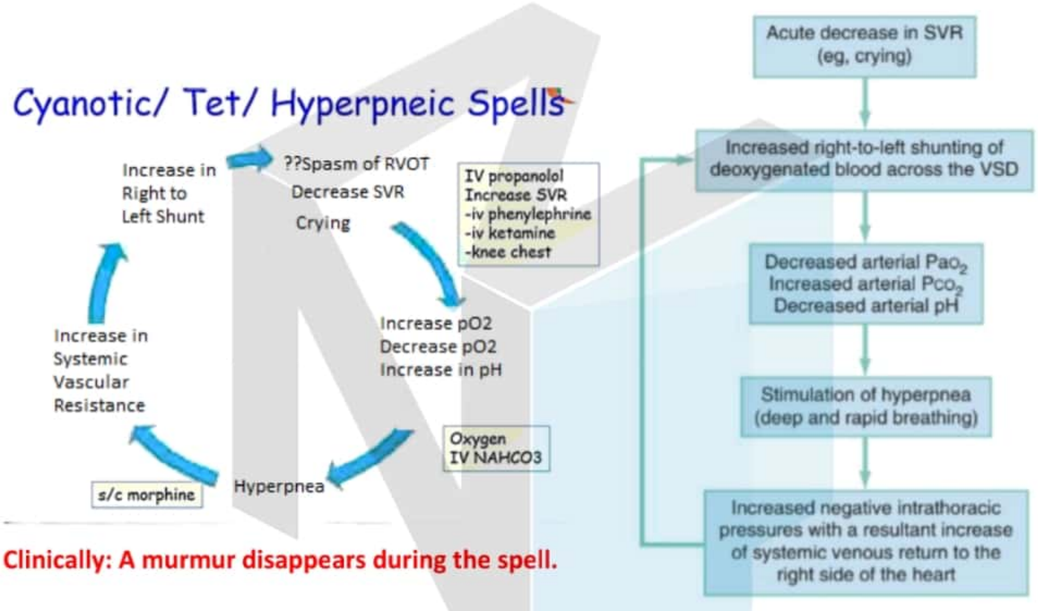
- **The Boot-shaped Heart sign**
  "coeur en sabot"
  - The UPPER part of the Boot: flat / concave pulmonary trunk
  - The TOE--upward pointing cardiac apex: concentric right ventricular hypertrophy

Accentuated by large lung volume, small thymus, and lordotic projection

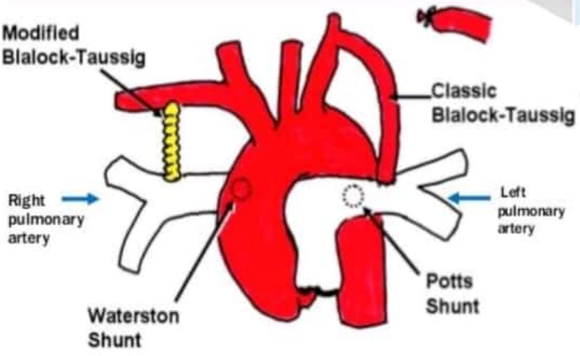
HyperCyanotic Spells/Tet Spells/HyperPneic Spells

Pathophysiology

1. *Event that suddenly lowers the SVR, such as crying or defecation, and hypovolemia or tachycardia will produce a large right-to-left shunt across the VSD, beginning the vicious circle of a hypoxic spell.*
2. *Shunt through the VSD bypasses the lungs and causes hypercarbia, hypoxemia, and acidosis*
3. *Respiratory centres are stimulated and the child hyperventilates*
4. *More negative intrathoracic pressure increases the amount of blood returning to the right side of the heart*
5. *The systemic blood shunts across the VSD leading to further hypoxia*



Clinically: A murmur disappears during the spell.



A classic Blalock-Taussig shunt (BT shunt) connects the right or left subclavian artery directly to the left or right pulmonary artery.

A modified BT shunt consists of a Gore-Tex graft that then connects the subclavian artery to the pulmonary artery. The advantage to a modified BT shunt is having more controlled blood flow.

A Waterston shunt connects the ascending aorta to the main or right pulmonary artery.

The Potts shunt connects the descending aorta to the pulmonary artery.

Dextro-Transposition of the Great Arteries/Vessels (d-TGA)

- M/C Cyanotic Heart Disease in newborns
- Two problems mainly:
  - Unoxygenated blood present in the systemic circulation
  - Oxygenated blood present in the lungs

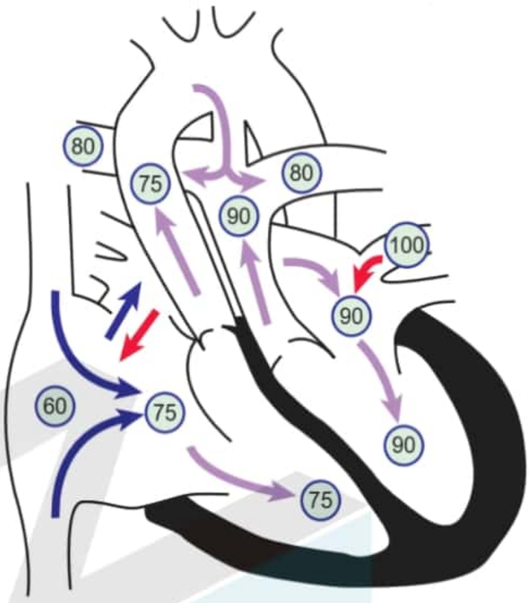


Figure 431-1 Physiology of d-transposition of the great arteries (d-TGA). *Circled numbers* represent oxygen saturation values. Right atrial (mixed venous) oxygen saturation is decreased secondary to systemic hypoxemia. Desaturated blood enters the right atrium, flows through the tricuspid valve into the right ventricle, and is ejected into the transposed aorta with resultant severe aortic desaturation. Fully saturated pulmonary venous blood flows into the left atrium, across the mitral valve into the left ventricle, and across the transposed pulmonary artery into the lungs. Pulmonary arterial oxygen saturation is thus increased. This lesion would not be compatible with life were it not for the ability of blood to shunt via 2 fetal pathways: the patent foramen ovale (PFO) and patent ductus arteriosus (PDA). Blood may shunt left to right or bidirectionally at the PFO. Because systemic vascular resistance tends to be higher than pulmonary vascular resistance, blood tends to shunt across the PDA mostly from the aorta to the pulmonary artery. As pulmonary resistance drops in the 1st few wk of life, pulmonary blood flow will gradually increase in patients with d-TGA.

- XRAY: Egg on string Appearance

- Treatment: When transposition is suspected, an infusion of prostaglandin E1 should be initiated immediately to maintain patency of the ductus arteriosus and improve oxygenation (dosage: 0.01-0.20 µg/kg/min). Because of the risk of apnea associated with prostaglandin infusion, an individual skilled in neonatal endotracheal intubation should be available.
- Infants who remain severely hypoxic or acidotic despite prostaglandin infusion should undergo **Rashkind balloon atrial septostomy**
- **The arterial switch (Jatene) procedure** is the **surgical treatment of choice for neonates** with d-TGA and an intact ventricular septum

Kawasaki Disease

C/F:

1. **Fever**
2. **Rash**
3. **Conjunctival Effusion**
4. **Swollen Palms and Soles**
5. **Desquamation**
6. **Strawberry Tongue**

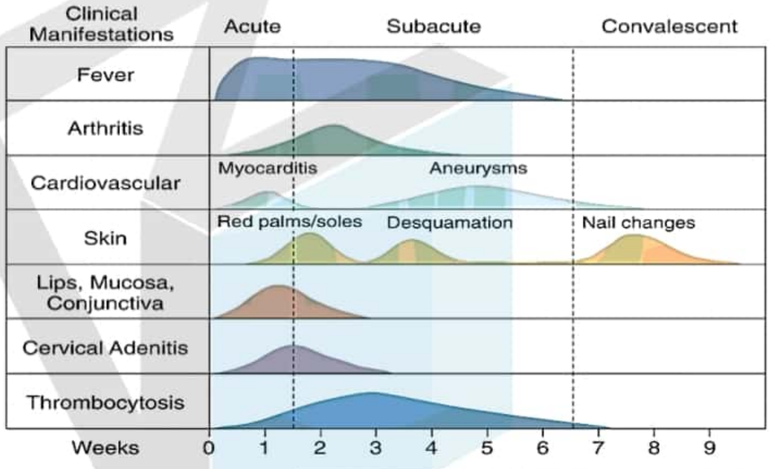
- Multisystem inflammatory syndrome in children (MIS-C) is a Kawasaki like illness, now said to be linked to **coronavirus** disease 2019 (**COVID**-19).

- Desquamation and Aneurysms appear in 2nd-3rd week of the illness.

- Most common acquired heart disease developed; US & JAPAN

- 20-25% Coronary artery aneurysm [2nd-3rd week]

- However this risk of formation of aneurysm is as less as 5% if timely IVIG therapy is given.



EPIDEMIOLOGIC CASE DEFINITION
(CLASSIC CLINICAL CRITERIA)*
Fever persisting at least 5 days†
Presence of at least 4 principal features:
Changes in extremities:
Acute: Erythema of palms, soles; edema of hands, feet
Subacute: Periungual peeling of fingers, toes in weeks 2 and 3
Polymorphous exanthem
Bilateral bulbar conjunctival injection without exudate
Changes in lips and oral cavity: erythema, lip cracking, strawberry tongue, diffuse injection of oral and pharyngeal mucosa
Cervical lymphadenopathy (>1.5 cm diameter), usually unilateral
Exclusion of other diseases with similar findings‡

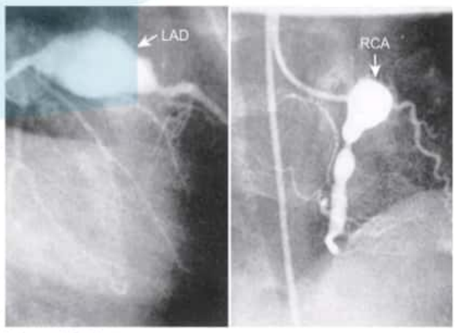


Figure 166-6 Coronary angiogram demonstrating giant aneurysm of the left anterior descending coronary artery (LAD) with obstruction and giant aneurysm of the right coronary artery (RCA) with an area of severe narrowing in 6 yr old boy. *(From Newburger JW, Takahashi M, Gerber MA, et al: Diagnosis, treatment, and long-term management of Kawasaki disease, Pediatrics 114:1708–1733, 2004.)*

- **Kobayashi score** is the most widely used and has high sensitivity and specificity.

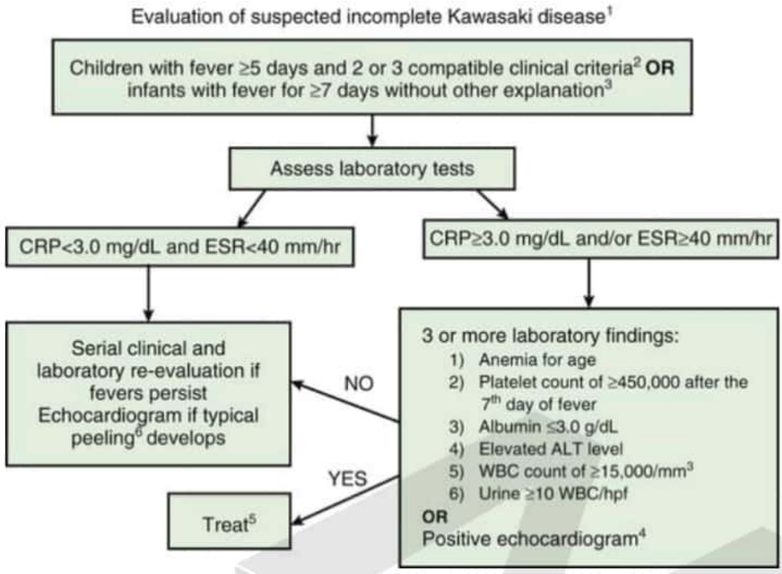

Evaluation of suspected incomplete Kawasaki disease[1]
Children with fever ≥5 days and 2 or 3 compatible clinical criteria[2] OR infants with fever for ≥7 days without other explanation[3]
Assess laboratory tests
CRP<3.0 mg/dL and ESR<40 mm/hr
CRP≥3.0 mg/dL and/or ESR≥40 mm/hr
Serial clinical and laboratory re-evaluation if fevers persist Echocardiogram if typical peeling[6] develops
NO
YES
Treat[5]
3 or more laboratory findings:
1) Anemia for age
2) Platelet count of ≥450,000 after the 7th day of fever
3) Albumin ≤3.0 g/dL
4) Elevated ALT level
5) WBC count of ≥15,000/mm3
6) Urine ≥10 WBC/hpf
OR
Positive echocardiogram[4]

Table 166-3 Treatment of Kawasaki Disease

ACUTE STAGE
- Intravenous immunoglobulin 2 g/kg over 10-12 hr *and*
- Aspirin 80-100 mg/kg/day divided every 6 hr orally until patient is afebrile for at least 48 hr

CONVALESCENT STAGE
- Aspirin 3-5 mg/kg once daily orally until 6-8 wk after illness onset if normal coronary findings throughout course

LONG-TERM THERAPY FOR PATIENTS WITH CORONARY ABNORMALITIES
- Aspirin 3-5 mg/kg once daily orally
- Clopidogrel 1 mg/kg/day (maximum: 75 mg/day)
- Most experts add warfarin or low-molecular-weight heparin for those patients at particularly high risk of thrombosis

ACUTE CORONARY THROMBOSIS
- Prompt fibrinolytic therapy with tissue plasminogen activator or other thrombolytic agent under supervision of a pediatric cardiologist

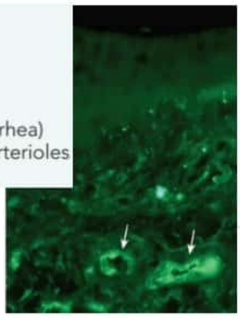
Henoch Schoenlein Purpura

- Most common vasculitis in children
- Most common small vessel vasculitis in children
- Most common Leucocytoclastic vasculitis in children
- a/k/a IgA Vasculitis

AMERICAN COLLEGE OF RHEUMATOLOGY CLASSIFICATION CRITERIA†

Two of the following criteria must be present:

- Palpable purpura
- Age at onset ≤20 yr
- Bowel angina (postprandial abdominal pain, bloody diarrhea)
- Biopsy demonstrating intramural granulocytes in small arterioles and/or venules



Skin biopsy of lesion from the same patient, showing direct immunofluorescence of immunoglobulin A within the walls of dermal capillaries.

HSP Nephritis is usually seen by 6weeks.

Renal followup can be done till 6 months

KIDNEYS- 50%

- 4 wks 84%
- 6 wks 91%
- 6 months 97%

SUPRAVENTRICULAR TACCHYCARDIA

- M/C Arrythmia in children

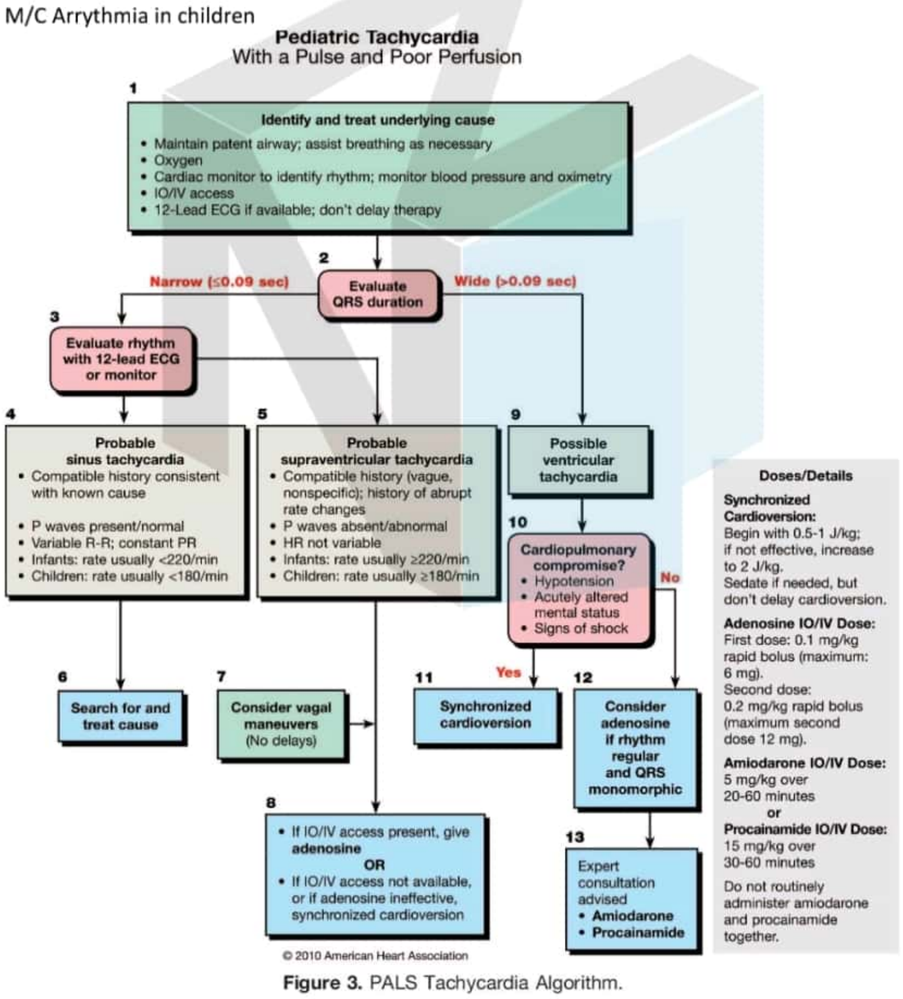

Pediatric Tachycardia
With a Pulse and Poor Perfusion
1
Identify and treat underlying cause
• Maintain patent airway; assist breathing as necessary
• Oxygen
• Cardiac monitor to identify rhythm; monitor blood pressure and oximetry
• IO/IV access
• 12-Lead ECG if available; don't delay therapy
2
Evaluate QRS duration
Narrow (≤0.09 sec)
Wide (>0.09 sec)
3
Evaluate rhythm with 12-lead ECG or monitor
4
Probable sinus tachycardia
• Compatible history consistent with known cause
• P waves present/normal
• Variable R-R; constant PR
• Infants: rate usually <220/min
• Children: rate usually <180/min
5
Probable supraventricular tachycardia
• Compatible history (vague, nonspecific); history of abrupt rate changes
• P waves absent/abnormal
• HR not variable
• Infants: rate usually ≥220/min
• Children: rate usually ≥180/min
9
Possible ventricular tachycardia
10
Cardiopulmonary compromise?
• Hypotension
• Acutely altered mental status
• Signs of shock
No
Yes
6
Search for and treat cause
7
Consider vagal maneuvers (No delays)
11
Synchronized cardioversion
12
Consider adenosine if rhythm regular and QRS monomorphic
8
• If IO/IV access present, give adenosine
OR
• If IO/IV access not available, or if adenosine ineffective, synchronized cardioversion
13
Expert consultation advised
• Amiodarone
• Procainamide
Doses/Details
Synchronized Cardioversion:
Begin with 0.5-1 J/kg; if not effective, increase to 2 J/kg.
Sedate if needed, but don't delay cardioversion.
Adenosine IO/IV Dose:
First dose: 0.1 mg/kg rapid bolus (maximum: 6 mg).
Second dose: 0.2 mg/kg rapid bolus (maximum second dose 12 mg).
Amiodarone IO/IV Dose:
5 mg/kg over 20-60 minutes
or
Procainamide IO/IV Dose:
15 mg/kg over 30-60 minutes
Do not routinely administer amiodarone and procainamide together.
© 2010 American Heart Association

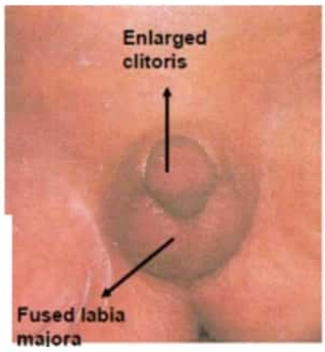
Figure 3. PALS Tachycardia Algorithm.

Parameter		Sinus tachycardia	SVT
Heart rate	Infants	Usually <220/min	Usually ≥220/min
	Children	Usually <180/min	Usually ≥180/min
History		Gradual onset, compatible with known cause e.g. fever, pain, volume loss.	Abrupt onset, vague, non-specific history, symptoms of CHF.
P-wave		Present/normal (upright in I/aVF)	Absent/abnormal (negative in leads II/III/aVF)
R-R interval		Variable with level of activity	Not variable
P-R interval		Constant	Variable
Chest Radiograph		Usually small heart and clear lungs unless pneumonia or underlying heart disease.	Signs of CHF (e.g. cardiomegaly, pulmonary edema) may present.

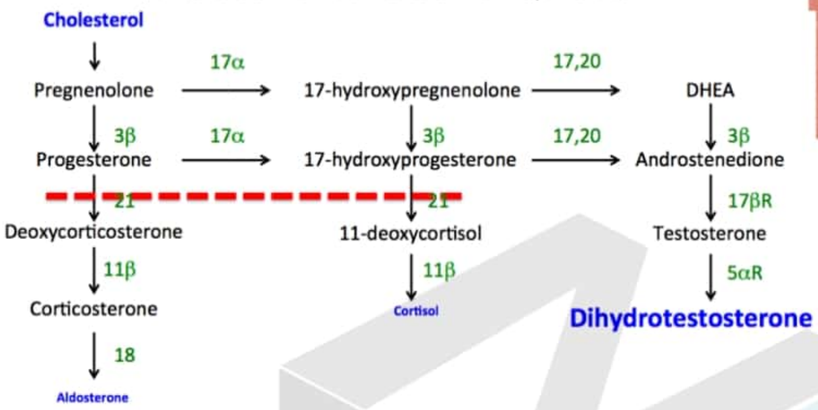
Child with Fever + Stridor

	Croup	Epiglottitis	Bacterial Tracheitis
Peak Age	6 months to 3 years (peak at 2-years-old)	3 to 7 years	3 to 8 years
Clinical presentation	URI symptoms with barky, seal-like cough, inspiratory stridor, low-grade fever	Rapid progression of high fever, drooling, stridor	URI prodrome similar to croup that intensifies to include high fever, inspiratory and expiratory stridor
Most common cause	• Parainfluenza virus	• *H. influenzae* • Strep sp. • *S. aureus*	• *S. aureus* • Mixed flora
X-ray findings	• Steeple sign on PA view	• Thumbprint sign on lateral neck view	• Subglottic narrowing • Ragged edge of tracheal air column • Hazy density within tracheal lumen
Clinical appearance	Non-toxic	Toxic	Toxic
Vaccine available	No	Yes (for *H. influenzae*)	No
Management	• Steroids • Aerosolized epinephrine	• Airway management (intubation) • Antibiotics	• Airway management (intubation) • Antibiotics

Congenital Adrenal Hyperplasia (CAH)



- Neonate in shock
- Has ambiguous genitalia
- Male: Phallus + undescended testis
- M/C Cause of Female Pseudo-Hermaphrodite



- Minimal formation of Cortisol
- Minimal formation of Aldosterone → Salt wasting Crisis
- Excess formation of Testosterone → Ambiguous Genitalia in Girls, Precocious Puberty in Boys
- Test of Choice: 17-alpha-hydroxyprogesterone (Neonatal Screening test)

CAH-One Liners:

- 90% 21-hydroxylase deficiency
- 70% present as salt wasting crisis

Q. Diagnostic modality for other biochemical diseases: TMS-Tandem mass Spectrometry

Treatment of CAH: Intravenous 10% Hyrdoxycortisone with NS + FLUDROCORTISONE

Table 1. Enzyme Deficiencies Resulting in CAH

Deficient Enzyme	Substrate	Product	Androgen	Mineralo-corticoid	% of CAH
21-hydroxylase	Progesterone	Deoxycorticosterone (DOC)	↑	↓	>90%
	17-hydroxy progesterone	11-deoxycortisol			
11β-hydroxylase	Deoxycorticosterone	Corticosterone	↑	↑	5%
Steroidogenic acute regulatory protein (STAR)	--	Necessary for cholesterol transport into mitochondria	↓	↓	Rare
3β-hydroxysteroid dehydrogenase (3β-HSD)	Pregnenolone, 17-OH pregnenolone, DHEA	Progesterone, 17-OHP, Δ^4-androstenedione	↓	↓	Rare
17α-hydroxylase	Pregnenolone	17-OH pregnenolone	↓	↑	Rare
	Progesterone	17-OH (17-OHP)			

SEVERE ACUTE MALNUTRITION (SAM)

1. **What is SAM?** → Weight/height<-3SD
2. **Acute malnutrition/ wasting?** → Weight/height
3. **Chronic malnutrition/ stunting** → Height/age (For both→ Weight/Age)
4. **Underweight?** →Weight/age<-2SD

Severe Acute Malnutrition (SAM)

This special classification is recommended by WHO for identifying and managing children with life threatening undernutrition in public health programme settings. Children with SAM have a mix of features of marasmus and kwashiorkor.

Severe acute malnutrition (SAM) among children 6–59 months of age is defined by WHO and UNICEF as any of the following three criteria:

i. Weight-for-height below –3 standard deviation (<–3SD) on the WHO Growth Standard; or
ii. Presence of bipedal edema; or
iii. Mid upper arm circumference (MUAC) below 11.5 cm.

In a child below 6 months of age, the MUAC is not used as a criterion.

NEPHROTIC SYNDROME

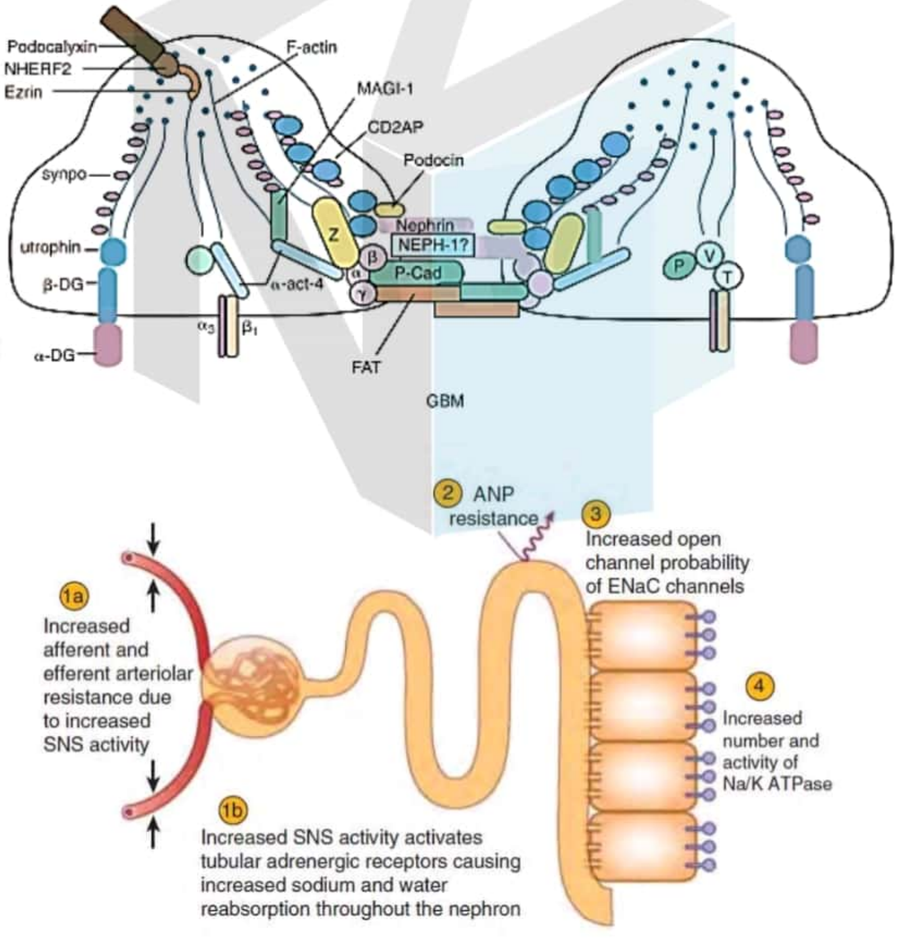


Table 527-3	Nephrotic Syndrome in Children Caused by Genetic Disorders of the Podocyte			
GENE	**NAME**	**LOCATION**	**INHERITANCE**	**RENAL DISEASE**
STEROID-RESISTANT NEPHROTIC SYNDROME				
NPHS1	Nephrin	19q13.1	Recessive	Finnish-type congenital nephrotic syndrome
NPHS2	Podocin	1q25	Recessive	FSGS
WT1	Wilms tumor-suppressor gene	11p13	Dominant	Denys-Drash syndrome with diffuse mesangial sclerosis Frasier syndrome with FSGS
LMX1B	LIM-homeodomain protein	9q34	Dominant	Nail-patella syndrome
SMARCAL1	SW1/SNF2-related, matrix-associated, actin-dependent regulator of chromatin, subfamily a-like 1	2q35	Recessive	Schimke immunoosseous dysplasia with FSGS*

DRUG OF CHOICE

- **Steroid resistant** Calcineurin inhibitors (TACROLIMUS > CYCLOSPORINE)
- **Steroid responsive**: - 1st step FRNS: levamisole;
  SDNS- Best Oral Cyclophosphamide x 12 weeks

(FRNS: Frequently Relapsing NS ; SDNS- Steroid Dependent NS)

For Finnish type Congenital NS: NEPHRECTOMY is the only treatment

MEDULLARY NEPHROCALCINOSIS

Causes:

- Prematurity
- Hypercalcemia
- Williams syndrome
- Primary neonatal hyperparathyroidism
- Paracellin 1 disorders, Bartter's syndrome
- Dent's disease; Lowe syndrome
- Cystinosis
- Distal RTA
- Calcium sensing receptor disorders
- Primary hyperoxaluria
- Cystic fibrosis

M/C/C of Medullary Calcinosis in Adults: Distal RTA

In premature babies: A/W Furosemide

Recurrent UTI

Protocol for Imaging of Recurrent UTI

1. USG
2. DMSA
3. MCU

1st UTI in a child →

- If < 1 yr- All above 3 tests (to rule out VUR)
- If b/w 1 to 5 years- USG, DMSA only. MCU only if any of these is abnormal
- If >5 years – USG only, DMSA only if USG is abnormal

Treatment of VUR

- **Patient less than 1 year or Grade 1 or 2 VUR→** Spontaneous Resolution as they age. Prophylaxis COTRIMOXAZOLE and wait & watch.
- **Patient more than 1 year and B/L Grade 3 or 4 VUR→** Surgery (Ureteric reimplantation)

Child with weak stream

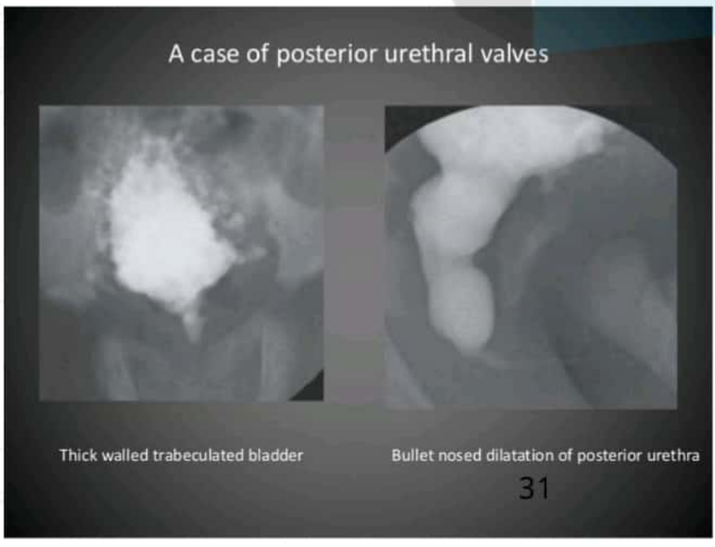
Probable Diagnosis: PUV (Posterior Urethral valve)

No obstruction when catheter is passed.

Obstruction present when child tries to micturate.

IOC: Micturating Cystourethrography (MCU) or VCUG

Rx: Cystoscopic Fulguration of the valve


A case of posterior urethral valves
Thick walled trabeculated bladder
Bullet nosed dilatation of posterior urethra
31